

## Human Chromosome Abnormalities: Analyzing Karyotypes

Take a look at the karyotype of a normal human male found on the back of this sheet. Notice he has 46 chromosomes. 22 of the pairs are considered autosomes, while the remaining pair, XY, are the sex chromosomes. The chromosomes can be arranged in seven groups (A-G) according to length. Group A consists of the six longest chromosomes. The B group consists of four long chromosomes with the centromeres very close to one end. The C group consists of 14 medium-length chromosomes with the centromeres slightly off-center. (The female sex chromosome (X) also falls into this group.) The D group consists of sex chromosomes slightly smaller than the C's with the centromeres very near one end. The E group resembles the C group, but the chromosomes are much smaller. The F group chromosomes are very small and have the centromeres in the middle. The G group includes the four smallest chromosomes with the centromeres so close to the ends that it is difficult to see any short arms at all. (The male sex chromosome (Y) falls into the G group.) Use this sample karyotype as a reference when completing the following activity.

You will be given a blank karyotype form and a biophoto sheet showing the chromosomes from an individual with an abnormal number of chromosomes. You need to create a completed karyotype revealing one of the following disorders:

- Trisomy 21 Down syndrome
  - Extra 21 chromosome
- Turner syndrome
  - Missing X chromosome
- Klinefelter syndrome
  - 2 X chromosomes AND a Y chromosome

First, count the chromosomes on the biophoto sheet and record this on your karyotype to help you narrow down your choices. Next, carefully cut out each of the individual chromosomes and arrange them on your blank karyotype form according to their size and number. Use the karyotype on the back of this sheet as a guide.

Note: Do NOT fasten the chromosomes to the karyotype form until you have arranged ALL of them and are sure of their position. Keep all paper scraps until you have identified each chromosome.

Once you are satisfied, glue down the chromosomes in the proper spaces. On the bottom of the completed karyotype, list the number of chromosomes, the sex of the subject, and the disorder according to the abnormality.